A previously asymptomatic two month-old infant with unique presentation of cor triatriatum sinister, right partial anomalous pulmonary venous return, and critical supravalvular stenosis of mitral valve

Dotychczas bezobjawowe dwumiesięcne niemowlę z unikatową prezentacją trójprzedsionkowego lewego serca, prawego częściowego nieprawidłowego spływu żył płucnych oraz krytycznej nadzastawkowej stenozy mitralnej

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Abstract

Major developments in diagnostic techniques in pre- and neonatal care have significantly reduced the rate of undetected congenital heart defects (CHD). Despite such advances, several patients with critical congenital heart defects are discharged annually from neonatal units with no proper diagnosis or treatment.

We present the case of a two month-old originally asymptomatic girl who represents the perfect example of such a situation. The infant was diagnosed just after pulmonary and cardiovascular decompensation with critical, complex CHD. The absence of disease symptoms of occurred due to a rare and specific morphology of pulmonary vessels and intracardiac membranes.

Key words: congenital heart defect, cor triatriatum sinister, anomalous pulmonary venous return, mitral stenosis, supravalvular, echocardiography, diagnosis, intervention, surgery, screening

Case report

The patient was born as a eutrophic neonate at the 38th week of gestation vaginally, reaching 10 in the Apgar score. The mother claimed it was her first pregnancy and she had received all recommended obstetrician ultrasonography (USG) examinations on time. The second trimester USG was repeated at the mother’s request. However, no defects were described in any of the prenatal examinations. The girl had no symptoms of cardiovascular disease. There was a negative result from the patients with critical congenital heart defects (CCHD) protocol screening pulse oximetry test. The first signs of a cardiac problem appeared in the second month of life, a few days after a scheduled vaccination. Slight fatigue and lack of appetite were noted, however no signs of infection or a gastrointestinal system problem were observed. The cautious mother decided to take the infant to see their general practitioner. Medical examination revealed no murmurs or objective signs of heart disease. Eventually, before leaving the doctor’s surgery, the child developed sudden peripheral cyanosis, consciousness disorders, and heavy dyspnoea. The patient was transported

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Shortly after, the patient underwent full surgical correction of pulmonary venous connections in conditions of extracorporeal circulation and deep hypothermia. The earlier diagnosis was finally confirmed intraoperatively as proper left pulmonary venous return was restricted by intraatrial structures blocking the mitral valve inflow. Additional membranes in the left atrium were resected with simultaneous restoration of the atrial septum with a pericardial patch. The overall condition of the patient soon after surgery was stable. Unfortunately, several post-operative complications occurred. The patient developed end-stage kidney failure, thus treatment with peritoneal dialysis was necessary as well as prolonged mechanical ventilation due to an inability to breathe unaided. Furthermore, various bacterial species were found in the blood tests, resulting in administration of a wide spectrum of antibiotics. A few days later, fungi were discovered in the patient’s urine which necessitated antifungal therapy. Due to these multiple health problems, total hospitalisation time was significantly prolonged and lasted up to two months after surgery.

Nevertheless, the patient survived and conquered all her challenges. At discharge, the girl was in an excellent condition and had no signs of her previous illnesses. ECHO revealed normalisation of chamber sizes and an optimal haemodynamic result of the surgical intervention. The six-month follow-up proved cardiologic management of the defect to be working, as the patient was developing correctly without any disabilities (Figures 1–3).

Figure 1. Preoperative state: A, B. Observable pressure and volume overload of right atrium and ventricle; C. Schematic presentation of concomitant defects.
Discussion

The presented case describes a very rare situation wherein a coexistence of several factors led to the misdiagnosis of a severely ill child. Early detection of CCHD is crucial as any delay in diagnosis significantly affects morbidity and mortality. Such early detection is possible in the antenatal period and is now common due to the implementation of the pulse oximetry screening test [1]. The examination result is based on the observable decreased blood saturation level in neonates with CCHD. The test is highly sensitive and specific. However, several reports of undetected serious cardiac defects appear annually [2]. The main limitation of the method is the differing time of transformation of the foetal circulation into a post-natal one, especially when a unique heart morphology co-exists. If the pulse oximetry test is performed before shunt closure, it can give a false negative result. Such a situation probably occurred in the presented case. The patient screening test went negatively despite the life-threatening anomaly. The presentation of symptoms in such a situation is typically a matter of time. Prolonged shutting of physiological shunts and enlarging obstruction of inflow usually leads to haemodynamic decompensation in infancy. The sudden aggravation of this patient’s condition appeared only after a rapid decrease in the amount of blood flowing into the left atrium, resulting in cardiac shock.

Nonetheless, standard post-natal pulse oximetry screening is not the only method to determine whether a child is in the high risk group for CHD existence. Obstetrician USG pre-natal screening is popular worldwide and provides moderately sensitive and specific data about the foetal heart and vessels [3]. The most common objection made against this method is that the variation in skill levels among USG operators determines the diagnostic outcome of the examination. If performed incautiously, or by a person not trained in foetal heart visualisation, CHD can easily be
missed [4]. Even so, rare or not prominent CHD may be described as a variant of a normal developing heart. This is probably true of the presented patient because double second trimester USGs (by two different operators) revealed no red flag symptoms requiring referral to the prenatal cardiology unit.

The difficulty level of diagnosing the presented patient was also increased due to a very uncommon and specific concomitance of several heart pathologies which may not give any symptoms in the first months of life. The prevalence of cor triatriatum sinister is estimated to be only 0.4% of all CHDs. It is best described as left atrium division into two compartments by an additional septum — the first with pulmonary venous return and the second with mitral inflow [5]. The presented patient was also complicated by horizontal configuration of abnormal membranes in the left atrium with trans-membranous obstructed flow resulting in severe supravalvular mitral stenosis (MS). Such a situation has only been described in a few cases [6]. Partial anomalous pulmonary venous return is a condition defined as abnormal connection of one-sided pulmonary veins to the incorrect atrial chamber or vessels. It is found in approximately 0.4–0.7% of all CHDs [7]. The case described here is cardiac type right PAPVR. However, in the first two months of the girl’s life, the small connection between the atrial chambers and the foramen ovale was probably persistent.

Unfortunately, the coexistence of all anomalies (CTS, MS and PAPVR), as well as heart growth with restricting flows, resulted in a very rare clinical presentation imitating total anomalous pulmonary venous return that affected the patient’s condition dramatically due to the critical limitation of inflow to the left ventricle and the massive overload of pulmonary circulation.

The essence of this case is a long chain of rare pathophysiologic events leading to a lack of diagnosis in the pre- and postnatal periods causing a life-threatening situation. It also shows the limitations of screening techniques, and thus emphasises the role of proper training for USG operators and the significance of a cautious neonatal examination based on experience and additional tests.

References